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Ophthalmic Genetics: A New Synthesis

AS INFECTIOUS DISEASES such as rubella have been brought under control with improved public health measures and immunizations, genetic factors have become increasingly important causes of both congenital and acquired eye diseases, particularly in the pediatric age group. Although accurate statistics are not available in the United States, it is estimated that at least 50% of new cases of legal blindness (20/200, or less than 20 degrees of peripheral vision) in the pediatric age group can be attributed to genetic causes; the economic and social consequences of this visual impairment are devastating. An ophthalmologist assumes a central role in identifying the cause of such visual handicaps and may assist pediatricians or geneticists in establishing a diagnosis in an otherwise perplexing patient. Referral is of particular importance in the detection of unilateral visual impairment, as a child with good vision in one eye may be asymptomatic. The eye is affected relatively early in the course of many genetic metabolic diseases and an ophthalmologist may be the first physician consulted. For example, patients who have Spielmeyer-Vogt disease, a lipopigment storage disorder, may have decreased central vision and ophthalmoscopic findings compatible with juvenile macular degeneration; progressive mental deterioration and visual impairment ensue. For some disorders, such as mannosidosis and Fabry's disease, the ocular manifestations are unique and diagnostic.

The hereditary bases of diseases that affect the eye include all three broad categories: chromosomal aberrations, single gene mutations consistent with Mendelian inheritance patterns and multifactorial inheritance. Some hereditary disorders that affect the eye have multiple genetic causes. The incidence of retinitis pigmentosa is about 1 in 3,000, much more common than would be expected from a disease caused by a single gene. Inheritance follows autosomal-dominant, autosomal-recessive and X-linked patterns. Although retinitis pigmentosa was thought to be a hereditary disease in all cases, a recent analysis of a large group of patients using segregation analysis, a complicated mathematical model, showed that some forms are not hereditary; the responsible environmental factors are not yet known.

The story of retinoblastoma has recently been unraveled. Strong evidence implicates a gene for this ocular malignancy on the short arm of chromosome 13 near the gene for esterase D, an enzyme that is unrelated to retinoblastoma. It is now believed that both the hereditary and the chromosomal forms of the disease are caused by an abnormality of the same gene. Neither the DNA sequence nor the biologic function of this oncogene has been identified.

The precedent for bringing a lawsuit for a wrongful birth has been set in the state of California by several cases that have been decided by the Supreme Court. Ophthalmologists and other physicians should consider the possibility of genetic factors in their visually handicapped patients; geneticists are well trained to relay information about hereditary disorders and congenital malformations without imposing value judgments.

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Laser Treatment for Glaucoma

THE OPHTHALMIC LASER was one of the earliest clinical applications of laser technology to medicine. Treatment of ocular disease primarily involved laser therapy for diseases of the macula, retinal tears and diabetic retinopathy. More recently, the argon laser has become a significant aid in the surgical treatment of both open-angle and closed-angle glaucoma. By using the laser, the number of patients requiring a conventional glaucoma operation has been reduced along with the associated complications, prolonged hospital care and inconvenience to patients and physicians.

Argon laser trabeculoplasty is the procedure used for open-angle glaucoma. Currently, it is the primary procedure of choice when the intraocular pressure cannot be adequately controlled medically. By placing small laser burns in the trabecular meshwork of the anterior chamber, the outflow facility of the eye increases and the intraocular pressure decreases. The 50 micron-sized burns are not full thickness, but cause stretching of the adjacent trabecular meshwork and enlarge the outflow channels. The surgical procedure is done on an outpatient basis, requires only topical anesthesia and is associated with minimal patient discomfort. Long-term follow-up is not available as the procedure first became a clinical research tool in the middle and late 1970s; however, three-year follow-up studies show an average decrease in intraocular pressure of 6 to 8 mm of mercury.

The optimal number of burns, their exact placement and the extent of the angle area to be treated are issues still under investigation.

For surgical management of cases of narrow (occludable) anterior chamber and narrow-angle glaucoma, argon laser iridotomy has replaced surgical iridectomy in all but the most difficult cases. This procedure is also done on an outpatient basis with topical anesthesia and minimal discomfort. In laser iridotomy, light is absorbed by the melanin pigment in the iris. The heat generated from the laser burn produces a